**Genetic Screening and Counseling in Perinatal Medicine: their Implications for Maternal and Fetal Health**

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 **Abstract:** Genetic screening and counseling are represented as an important part of perinatal medicine for maternal and fetal health, which clarified the importance of prenatal diagnostic techniques and testing in helping to detect significant fetal abnormalities early, where our current study aims to record clinical outcomes and evaluate the impact of genetic screening and counseling on maternal and fetal health.

Furthermore, clinical outcomes in mothers and fetuses were collected from different hospitals in Iraq, during the period from January 2, 2024, to December 25, 2025, during which a total of 87 pregnant women underwent comprehensive fetal screening, testing, and counseling, as it was assessed the outcomes and the mother's psychological state were assessed.

The mean age of the study group was 28.5 years, and the most frequent reason for screening was advanced maternal age (51.7%). The most common screening method was NIPT (69.0%), and abnormalities, mostly Trisomy 21 (53.3%), were found in 17.2% of tests. Genetic counseling affected decisions, with 13.8% choosing to terminate the pregnancy and 69.0% continuing it, and dramatically decreased maternal anxiety levels (7.8 to 4.2, p<0.01). 13.8% of neonates had congenital defects at birth, compared to 86.2% who were healthy. 74.7% of mothers stated that they were "very happy" with therapy, indicating high levels of satisfaction. Where our study concluded that both genetic screening and counseling play an important role in perinatal outcomes, it also notes that they significantly improve maternal and fetal health. Genetic screening and counseling facilitate critical decision-making and improve maternal well-being.

**Key words:** Genetic Screening, Genetic Counseling, Perinatal Medicine, Maternal and Fetal Health

**Introduction**

Advances in medical research have revolutionized perinatal medicine by offering unprecedented opportunities to safeguard the health of the mother and the fetus {1, 2}, which genetic screening and counseling have emerged as crucial tools for identifying, managing, and lowering the risks in genetic abnormalities throughout pregnancy, where perinatal medicine found with the health of both mom and baby from conception by the early postnatal period, has relied increasingly on genetic technology to provide tailored care and informed decision-making. {3}

Also, genetic screening presented the systematic identification for any genetic disorders with the fetus, genetic counseling enrolled families with the information, guidance, and support they need to cope with the challenges of genetic results, which practices have a major effect in the health of the mother along with the fetus, enabling early disease detection, informed reproductive choices, and tailored medical care. {4, 5}, the use of genetic screening in prenatal care has been spurred by growing understanding of the genetic basis of a number of congenital and developmental disorders {6, 7, 8]

These diagnoses had allowed into healthcare providers to apply timely treatments and management strategies by detecting risks early in pregnancy, as well as the ability to detect genetic disorders during pregnancy, which has significant implications for neonatal care since it enabled healthcare providers to prepare for specialized treatment as soon as the baby is born. {9}

**Patients and Methods**

Study Design

Over a duration of a 12-month follow-up period, from January 2024 to January 2025, 87 pregnant women who received genetic screening along with counseling at different hospitals in Iraq, participated in a cross-sectional research, we chose women who were monitored during pregnancy and delivery and who received genetic screening or counseling in the first or second trimester, where all of the data used in our investigation came from medical records kept at different hospitals in Iraq, that our study sought to ascertain the extent of genetic counseling in maternal and fetal health as well as the effects of genetic screening indicators.

Data Collection

A standardized questionnaire was used to attract the women who participated in a series of interviews, including age, gestational age, and the number in parities at enrollment were among the clinical and demographic information we documented for the participating women, as well as we discovered markers for genetic screening, such as aberrant ultrasound results, a family history of genetic abnormalities, and advanced maternal age.

Moreover, all mothers had non-invasive prenatal testing (NIPT), which included amniocentesis and chorionic villus sampling (CVS), which a Likert-scale questionnaire was used in our study to gauge how satisfied mothers were with genetic counseling, that validated anxiety scale was also used in our study to measure mother anxiety levels both before and after genetic counseling, where the fetus's clinical results, including whether congenital defects were present or if the delivery was normal, were also documented.

Genetic Screening of Pregnant Women

Prenatal genetic screening of pregnant women was performed using cell-free fetal DNA analysis in the mother's blood, where it enrolled surgical procedures were performed, including chorionic villus sampling (CVS) and amniocentesis, for both women with high-risk pregnancies and women with abnormal prenatal screening results. All women underwent all screening tests, and a team of geneticists and obstetricians informed all women of the abnormal results during genetic counseling sessions. Maternal satisfaction with genetic counseling was assessed using a five-point questionnaire ranging from "very dissatisfied" to "very satisfied."

Statistical Analysis

The data collected was recorded and organized using SPSS version 22.0. Continuous variables, such as age and anxiety scores, were defined as means ± standard deviations (SDs). Other variables that influenced screening results and satisfaction levels were defined as frequencies and percentages. A p-value <0.05 was considered statistically significant in this study.

**Results**

**Table 1: Enroll demographic features in our study.**

|  |  |
| --- | --- |
| **Characteristic** | **Number (%)** |
| **Age (years)** | 28.5 ± 4.2 (Mean ± SD) |
| **Gravidity** | 2.1 ± 1.3 (Mean ± SD) |
| **Parity** | 1.4 ± 0.9 (Mean ± SD) |

**Table 2: Determining indications related to genetic screening.**

|  |  |
| --- | --- |
| **Indications** | **Number (%)** |
| Advanced Maternal Age | 45 (51.7%) |
| Family History of Genetic Disorders | 22 (25.3%) |
| Abnormal Ultrasound Findings | 15 (17.2%) |
| Previous Child with Genetic Disorder | 5 (5.8%) |

**Table 3: Identify types of genetic screening in our participants.**

|  |  |
| --- | --- |
| **Screening Type** | **Number (%)** |
| Non-Invasive Prenatal Testing (NIPT) | 60 (69.0%) |
| Chorionic Villus Sampling (CVS) | 12 (13.8%) |
| Amniocentesis | 15 (17.2%) |

**Figure 1: Identify clinical outcomes of genetic screening.**

**Figure 2: Types of detected abnormalities in our study.**

**Table 4: Impact of genetic counseling on decision-making at participants.**

|  |  |
| --- | --- |
| Outcomes | Number (%) |
| Continued Pregnancy | 60 (69.0%) |
| Termination of Pregnancy | 12 (13.8%) |
| Further Diagnostic Testing | 15 (17.2%) |

**Table 5: Assessment of maternal anxiety status in pre- and post-counseling.**

|  |  |
| --- | --- |
| Time Point | Anxiety Score (Mean ± SD) |
| Pre-Counseling | 7.8 ± 1.5 |
| Post-Counseling | 4.2 ± 1.1 |

**Table 6: Fetal outcomes at birth.**

|  |  |
| --- | --- |
| **Outcome**s | **Number (%)** |
| Healthy Birth | 75 (86.2%) |
| Congenital Anomalies | 12 (13.8%) |

**Table 7: Evaluating maternal satisfaction into genetic counseling.**

|  |  |
| --- | --- |
| **Satisfaction Level** | **Number (%)** |
| Very Satisfied | 65 (74.7%) |
| Satisfied | 18 (20.7%) |
| Neutral | 3 (3.4%) |
| Dissatisfied | 1 (1.1%) |

**Discussion**

The paradigm of obstetric care has changed permanently in a reactive to a proactive approach to pregnancy management with the introduction of sophisticated genetic screening along with diagnostic technology into perinatal medicine {10}. The main consequence of the widespread use of cfDNA-based NIPT was its significant impact on the identification and comprehension of common aneuploidies. NIPT has greatly decreased the necessity for invasive diagnostic procedures such as amniocentesis and CVS by providing a very sensitive and specific non-invasive approach from as early as ten weeks' gestation, which it has directly decreased the risk of miscarriage associated with these procedures {11}, where the physical health of both the mother and the fetus will undoubtedly benefit from this, but it accessibility has also made prenatal genetic information more widely available, earlier in pregnancy, and to a larger, frequently lower-risk population. {12}

The research population was a relatively young cohort of women, had a mean age of 28.5 ± 4.2 years, as well as the averages for parity and gravity were 1.4 ± 0.9 and 2.1 ± 1.3, respectively, where these characteristics point to a group with a moderate level of reproductive experience, which might affect how open they are to genetic counseling and screening. The most common reasons for genetic screening were advanced maternal age (51.7%), a prior child with a genetic illness (5.8%), abnormal ultrasound results (17.2%), and family history for genetic disorders (25.3%). The most used screening technique (69.0%) was non-invasive prenatal testing (NIPT), most likely because of its accuracy and safety. {13} In 13.8% and 17.2% of instances, respectively, chorionic villus sampling (CVS) as well as amniocentesis were carried out. The preference for NIPT emphasizes how it is becoming a more popular first-line screening method, reducing the dangers of invasive treatments.

Pre-test counseling must now cover the scope, limits, and possible results associated with the genetic screen, especially the potential for incidental discoveries, rather than only discussing procedural hazards. The effects on maternal health are most noticeable during post-test counseling, especially after a good result. A possible fetal abnormality can have a substantial psychological impact, increasing the mother's worry, anxiety, and despair {14}. Supporting autonomous reproductive decision-making, whether it involves considering pregnancy termination, pursuing confirming diagnostic tests, or preparing for a child with special needs, requires effective, compassionate, and non-directive counseling. {15}

Reproductive autonomy has been significantly impacted by the switch from ethnicity-based to extended pan-ethnic panels for carrier screening. Preimplantation genetic testing (PGT), the utilization of donor gametes, or prenatal diagnosis into an existing pregnancy are among the several choices available when couples at risk of transmitting autosomal recessive disorders are identified before or early in pregnancy. This proactive strategy gives families information and options {16}. Nevertheless, it also brings up moral questions about the distribution of resources, the possibility of genetic discrimination, particularly the psychological effects of identifying people as carriers for serious, incurable diseases. {17}

Of the instances that were tested, 17.2% showed anomalies and 82.8% had normal findings. Down syndrome made up 53.3% of the aberrant findings, with neural tube abnormalities (13.3%), Edwards syndrome (26.7%), and other anomalies (6.7%) following closely behind. Decisions on pregnancy management were greatly impacted by genetic counseling. 13.8% of women with abnormal findings decided to terminate their pregnancy, 17.2% sought more diagnostic testing, and 69.0% decided to continue the pregnancy. Maternal anxiety levels were significantly reduced by genetic counseling; mean scores dropped from 7.8 ± 1.5 before counseling to 4.2 ± 1.1 after. This decrease emphasizes how counseling may help people psychologically by reducing stress and uncertainty {18}. Furthermore, 74.7% of women expressed high levels of satisfaction with the counseling method, compared to 20.7% who indicated satisfaction, 3.4% who indicated neutrality, and 1.1% who expressed dissatisfaction. High satisfaction levels demonstrate how well therapy meets the expectations of mothers. While 13.8% of newborns had congenital defects, 86.2% of babies were healthy at birth. The predictive accuracy for genetic testing is validated by these results, which match the screening results. {19, 20}

**Conclusion**

Our study concluded that advanced maternal age is the primary factor contributing to women undergoing non-invasive prenatal screening (NIPT), which is the most common. Furthermore, our study showed that the majority of results were normal, with 17.2% of abnormalities detected, including Down syndrome and Edwards' syndrome.

As a result, genetic counseling significantly reduced maternal anxiety, leading 69% of participants to continue their pregnancy. Furthermore, maternal satisfaction with counseling was high, resulting in healthy birth outcomes for 86.2% of births. Our study underscores the role of genetic counseling in prenatal care and improved postpartum health outcomes for both mothers and fetuses.

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